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Arthroplasty in patients with rare conditions

Total knee arthroplasty in ochronosis

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ABSTRACT

Alkaptonuria is disorder of tyrosine metabolism due to deficiency of homogentisic oxidase characterized by excretion of homogentisic acid in urine, deposition of oxidized homogensitate pigments in connective tissues and articular cartilages (ochronosis). The result is dark pigmentation and weakening of the tissues resulting in chronic inflammation and osteoarthritis. Management of alkaptonuric ochronic osteoarthritis is usually symptomatic and replacements have comparable outcomes to osteoarthritis in patients without ochronosis. I report a case of a patient with ochronosis of knee treated with total knee replacement and report operative pearls for surgery in this rare disorder.

Introduction

Alkaptonuria is an autosomal recessive disorder caused by the deficiency of homogentisate 1,2 dioxygenase activity (Fig. 1). It was first described by Virchow in 1866 [10]. Alkaptonuria is disorder of tyrosine metabolism due to deficiency of homogentisic oxidase characterized by excretion of homogentisic acid in urine, deposition of oxidized homogensitate pigments in connective tissues, the dermis, apocrine glands and articular cartilages (ochronosis). The pathogenesis of the disease is the polymerization of deposited HGA that discolors and weakens the connective tissue, ultimately resulting in brittle tissue that is easily disrupted and leads to chronic inflammation, degeneration, and eventually osteoarthritis [1]. Patients with alkaptonuria are usually asymptomatic and arthropathy appears after the fourth decade [2].

Systemically, there is thickening and blue–black or gray–blue discoloration of ear cartilage. Other body locations include the eyelids, sclera, foreheads, cheeks, axillae, genitals, nail beds, buccal mucosa, larynx, tympanic eardrum and nasal tip. Simultaneously, this discoloration can occur in skin, tendons, ligaments, costochondral junctions, sclera, heart valves, the intima of blood vessels and cause lumbar intervertebral disc calcification and disc space narrowing [3–5].

The knee is the most common site of orthopedic abnormality. Other sites of involvement are hips, shoulders, sacroiliac joints and the pubic symphysis [6]. There is currently no definitive cure for alkaptonuric ochronosis. However, total joint replacement in published cases of ochronotic osteoarthritis report good results similar to osteoarthritic patients without ochronosis [7] I report the case of a 58-year-old female with a family history of ochronosis, who developed degenerative arthritis of the knee.

Case history

A 58-year old female patient was admitted with history of bilateral knee pain for 8 years. Pain was of gradual onset, interfering her day to day activities, more on right side and was not responding to conservative treatment. No cutaneous signs of ochronosis were noted at time of presentation. The patient had a family history of ochronosis in her mother and 2 sisters. The patient had history of darkening of urine which was revealed by patient after surgery.

On local examination of right knee, there was no swelling. On deep palpation, there was medial and lateral joint line tenderness. The range of motion of the right knee was 0°–95° and left knee was 0°–105° with significant pain. The patient walked with an antalgic gait. Clinical diagnosis of osteoarthritis was confirmed with anteroposterior and lateral radiographs demonstrating joint spaces

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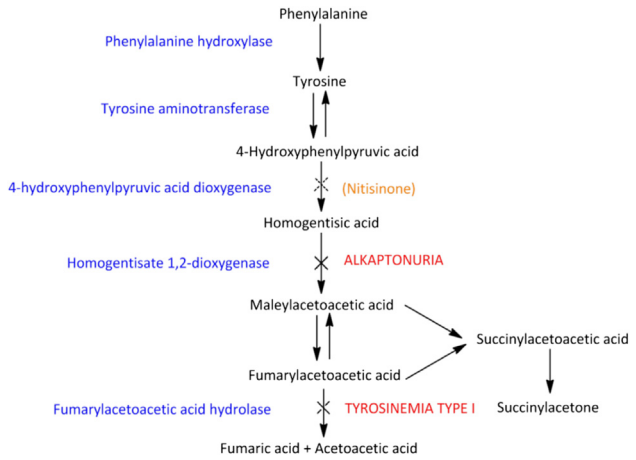

Figure 1. Diagram showing how phenylalanine is metabolized demonstrating how homogentisate 1,2 dioxygenase deficiency causes accumulation of homogentisic acid and causes alkaptonuria.

narrowing, peripheral new bone formation, subchondral bone sclerosis and arthritic changes of patella (Fig. 2). Cemented total knee replacement was offered after informed consent.

During surgery, an anterior midline incision was used. The subcutaneous tissues revealed black nodules (Figs. 3 and 4). There was synovial hypertrophy. The joint capsule was contracted and partially black with a hard consistency. It was difficult to retract the patella as patellar tendon was stiff and there was a popping sound during exposure. No obvious extensor mechanism dissociation was found. On capsular incision, I observed no joint fluid, blackened articular cartilage of femur, tibia and patella (Fig. 5) along with blackened undersurface of patella (Fig. 6) and quadriceps tendons. After total synovectomy, I noted dark black stiff menisci inside the joint (Fig. 5). While taking bone cuts black discoloration was seen up to the thickness of cartilage only and subchondral bone was of normal appearance but soft in consistency (Fig. 7). A $5 \times 3 \times 2$ mm cavitary defect was found in tibia which was ultimately filled by cement. Multiple drill holes were made on the raw surface of femur and tibia before cementing. Standard cementing technique was performed without difficulty (Fig. 8).

There was excessive bleeding during surgery interfering the operative field even after tourniquet control (300 mm Hg). There

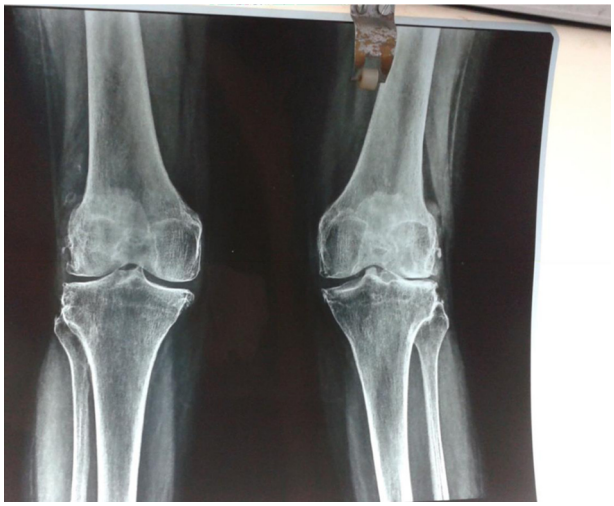

Figure 2. Pre-operative anteroposterior and lateral radiographs of the right knee suggesting features of osteoarthritis.

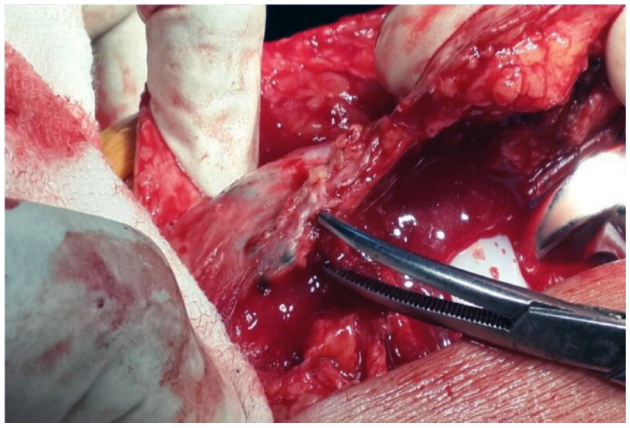

Figure 3. Subcutaneous nodules observed during surgery.

were no other major complications occurred during the surgery. The patient had increased blood loss postoperatively, which was totaled 1150 ml. The drain was removed on 4th postoperative day, which was atypical, but left in place because of blood loss. Patient had mild postoperative pain. Her operated joint was mobilized on second post-operative day. Post-operative wound healing was uneventful. Patient was very happy and satisfied with improved R.O.M. (0–110) and painless knee. The knee society score was 84 on last follow up 18 months post-operatively.

Discussion

Alkaptonuria is disorder of tyrosine metabolism due to deficiency of homogentisic oxidase characterized by excretion of homogentisic acid in urine, deposition of oxidized homogensitate pigments in connective tissues and articular cartilages (ochronosis). There is no definitive cure for the disease but Nitisinone, an inhibitor of 4-hydroxyphenyl pyruvate dioxegenase has been shown to decrease urinary excretion of homogentisic acid [11]. The effectiveness of Nitisinone in treating ochronosis is unknown.

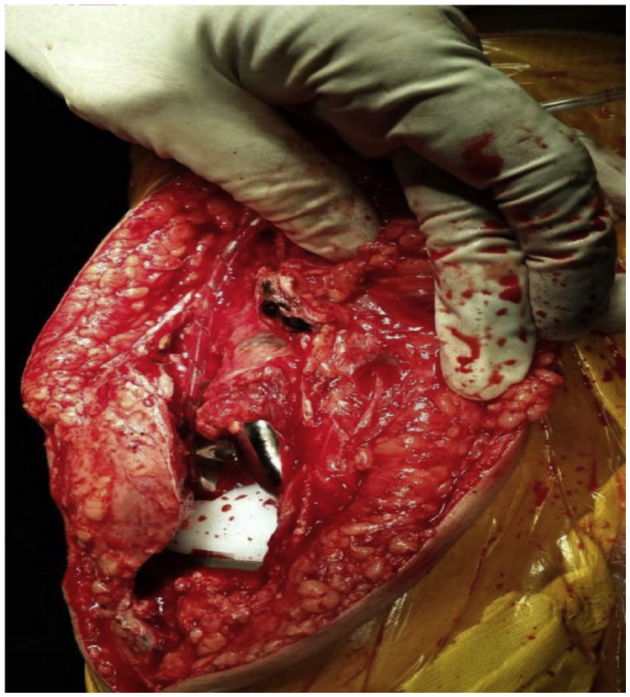

Figure 4. Soft tissue involvement in ochronosis.